Three Faces of Fragile X Syndrome

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### THREE FACES OF FRAGILE X SYNDROME

**Fragile X (FXS)** - Fragile X primary ovarian insufficiency (FXPOI) - Fragile X tremor/ataxia syndrome (FXTAS)

Fragile X is associated with a change to the *FMR1* gene of the chromosome Xq27.3. This single *FMR1* gene change will result in Fragile X-associated Disorders which includes three disorders that can cover multi generations in one family. PT interventions must vary accordingly.

#### FRAJGILE X

<table>
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<tr>
<th>&gt;200 CGG Repeats in the <em>FMR1</em> gene</th>
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<tbody>
<tr>
<td>Full FXS transmitted by mother to ☉ or ☽ (milder)</td>
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<td>All offspring from a FXS males will be typical</td>
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### Body structure Impairments

- Physical Impairments
  - Low Muscle tone
  - Hyperextensible joints
  - Mitral valve collapse
  - 2nd to abnormal elastin
  - High arch palate
  - Pectus excavatum
  - Soft skin
- Autistic like behavior with decreased eye contact

### Behavioral Impairments

- Functional Impairments
  - Delayed development (motor and speech)
  - Hyper/hypotonia sensory responses
  - Hyperactivity
  - Dislike change
  - Increased anxiety
- Social anxiety to point of depression or extreme shyness and/or social ineptness
- ADHD

### FRX PRIMARY OVARIAN INSUFFICIENCY

55-200 CGG Repeats in the *FMR1* gene (premutation range)

Transmitted from female carrier to 50% of female offspring who are then carriers

### Impairments in Childhood and Adolescence

- Premature menopause (≤5 years earlier than non-carrier) related to Primary ovarian insufficiency
- Early onset osteopenia progressing to osteoporosis
- Coronary heart disease
- Thyroid dysfunction
- Fibromyalgia and/or undifferentiated muscle pain
- Anxiety disorder
- Monitor at ±50 years for symptoms of FXTAS

### Activity Limitations

- Limited endurance, consider FXPOI as a basis

#### FXTAS FRX TREMOR/ ATAXIA SYNDROME

55-200 CGG Repeats in the *FMR1* gene (premutation range)

Transmitted from male parent to all daughters but not to sons, daughters will be carriers

### Body structure Impairments

- ≥50 years on-set of action/intention tremor and cerebellar ataxia or parkinsonism
- Cerebellar atrophy and increased T2 signal intensity in the middle cerebellar peduncles
- Balance problems with ataxia
- Cognitive decline, poor judgment and mood instability
- Numbness of the extremities, neuropathy

### Activity Limitations

- Gait instability with increasing falls
- Decreasing ability to carry out ADL skills
- Difficulty executing job responsibilities
- Decreased sexual function
- Driver safety risk with declining safety awareness

### PHYSICAL THERAPY IMPLICATIONS ~The Therapist’s primary task: to participate in the differential diagnosis process. The Therapist’s secondary task: to obtain a complete family history which should include the question: is there a family history of intellectual disabilities or autism spectrum disorders. This leads to the following interventions:

- Support developmental progress and postural control
- Biomechanical management for joint instability
- Develop, implement and monitor ongoing exercise program
- Develop ability to participate in peer activities
- Treat presenting impairment(s)
- Teach osteoporosis prevention and management
- Support implementing a regular exercise routine
- Differentiate normal aging from development of FXTAS
- Differentiate from other movement disorders i.e. parkinsonism
- PT identify and address cerebellar dysfunction, classify the ataxia
- Functional training to address limitations
- PT support to identify and implement adaptations

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